

Markus Schuelke

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

171 papers	15,530 citations	48 h-index	124 g-index
185 ext. papers	17,743 ext. citations	8.2 avg, IF	6.75 L-index

#	Paper	IF	Citations
171	Generation of four iPSC lines from four patients with Leigh syndrome carrying homoplasmic mutations m.8993T>G or m.8993T>C in the mitochondrial gene MT-ATP6.. <i>Stem Cell Research</i> , 2022 , 61, 102742	1.6	
170	What can go wrong in the non-coding genome and how to interpret whole genome sequencing data. <i>Medizinische Genetik</i> , 2021 , 33, 121-131	0.5	1
169	Mutation detection in the non-coding genome. <i>Medizinische Genetik</i> , 2021 , 33, 119-120	0.5	
168	Public data sources for regulatory genomic features. <i>Medizinische Genetik</i> , 2021 , 33, 167-177	0.5	1
167	Novel bi-allelic variants expand the SPTBN4-related genetic and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2021 , 29, 1121-1128	5.3	1
166	Defective metabolic programming impairs early neuronal morphogenesis in neural cultures and an organoid model of Leigh syndrome. <i>Nature Communications</i> , 2021 , 12, 1929	17.4	17
165	MutationTaster2021. <i>Nucleic Acids Research</i> , 2021 , 49, W446-W451	20.1	14
164	Successful plasmapheresis and immunoglobulin treatment for severe lipid storage myopathy: Doing the right thing for the wrong reason. <i>Neuropathology and Applied Neurobiology</i> , 2021 ,	5.2	0
163	Inflammation, fibrosis and skeletal muscle regeneration in LGMDR9 are orchestrated by macrophages. <i>Neuropathology and Applied Neurobiology</i> , 2021 , 47, 856-866	5.2	2
162	A New Mechanism in THRA Resistance: The First Disease-Associated Variant Leading to an Increased Inhibitory Function of THRA2. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	0
161	Extracellular matrix remodelling is associated with muscle force increase in overloaded mouse plantaris muscle. <i>Neuropathology and Applied Neurobiology</i> , 2021 , 47, 218-235	5.2	6
160	A novel mutation in NEB causing foetal nemaline myopathy with arthrogryposis during early gestation. <i>Neuromuscular Disorders</i> , 2021 , 31, 239-245	2.9	0
159	Expanding the clinical and molecular spectrum of ATP6V1A related metabolic cutis laxa. <i>Journal of Inherited Metabolic Disease</i> , 2021 , 44, 972-986	5.4	2
158	Diagnosis of Taenia solium infections based on "mail order" RNA-sequencing of single tapeworm egg isolates from stool samples. <i>PLoS Neglected Tropical Diseases</i> , 2021 , 15, e0009787	4.8	
157	Complement deposition at the neuromuscular junction in seronegative myasthenia gravis. <i>Acta Neuropathologica</i> , 2020 , 139, 1119-1122	14.3	8
156	Live-imaging of revertant and therapeutically restored dystrophin in the Dmd mouse model for Duchenne muscular dystrophy. <i>Neuropathology and Applied Neurobiology</i> , 2020 , 46, 602-614	5.2	3
155	Homozygous mutation in murine retrovirus integration site 1 gene associated with a non-syndromic form of isolated familial achalasia. <i>Neurogastroenterology and Motility</i> , 2020 , 32, e13923	4	0

154	Allan-Herndon-Dudley-Syndrome: Considerations about the Brain Phenotype with Implications for Treatment Strategies. <i>Experimental and Clinical Endocrinology and Diabetes</i> , 2020 , 128, 414-422	2.3	4
153	A spontaneous missense mutation in the chromodomain helicase DNA-binding protein 8 (CHD8) gene: a novel association with congenital myasthenic syndrome. <i>Neuropathology and Applied Neurobiology</i> , 2020 , 46, 588-601	5.2	2
152	Bi-Allelic UQCERS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. <i>American Journal of Human Genetics</i> , 2020 , 106, 102-111	11	19
151	A novel homozygous nonsense mutation of VPS13B associated with previously unreported features of Cohen syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 570-575	2.5	7
150	Presence of anti-neuronal antibodies in children with neurological disorders beyond encephalitis. <i>European Journal of Paediatric Neurology</i> , 2020 , 28, 159-166	3.8	1
149	A new homozygous HERC1 gain-of-function variant in MDFPMR syndrome leads to mTORC1 hyperactivation and reduced autophagy during cell catabolism. <i>Molecular Genetics and Metabolism</i> , 2020 , 131, 126-134	3.7	2
148	Defining the ATPome reveals cross-optimization of metabolic pathways. <i>Nature Communications</i> , 2020 , 11, 4319	17.4	3
147	Hybrid genome assembly and annotation of <i>Danionella translucida</i> . <i>Scientific Data</i> , 2019 , 6, 156	8.2	16
146	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. <i>Brain</i> , 2019 , 142, 2948-2964	11.2	13
145	MutationDistiller: user-driven identification of pathogenic DNA variants. <i>Nucleic Acids Research</i> , 2019 , 47, W114-W120	20.1	22
144	Muscle Weakness, Cardiomyopathy, and L-2-Hydroxyglutaric Aciduria Associated with a Novel Recessive SLC25A4 Mutation. <i>JIMD Reports</i> , 2019 , 43, 27-35	1.9	5
143	Fulminant cerebral venous thrombosis associated with the m.3243A>G MELAS mutation: A new guise for an old disease. <i>Brain and Development</i> , 2019 , 41, 901-904	2.2	1
142	RegulationSpotter: annotation and interpretation of extratranscriptomic DNA variants. <i>Nucleic Acids Research</i> , 2019 , 47, W106-W113	20.1	9
141	Human muscle-derived CLEC14A-positive cells regenerate muscle independent of PAX7. <i>Nature Communications</i> , 2019 , 10, 5776	17.4	21
140	Phenotero: Annotate as you write. <i>Clinical Genetics</i> , 2019 , 95, 287-292	4	3
139	Motor function in survivors of pediatric acute lymphoblastic leukemia treated with chemotherapy-only. <i>European Journal of Paediatric Neurology</i> , 2019 , 23, 304-316	3.8	5
138	De novo mutation in causes ichthyosis, , hypomyelination, spastic paraplegia, high frequency deafness and optic atrophy. <i>Journal of Medical Genetics</i> , 2019 , 56, 164-175	5.8	35
137	Caveolin 1 Promotes Renal Water and Salt Reabsorption. <i>Scientific Reports</i> , 2018 , 8, 545	4.9	6

136	Morvan syndrome associated with CASPR2 and LGI1 antibodies in a child. <i>Neurology</i> , 2018 , 90, 183-185	6.5	14
135	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. <i>Orphanet Journal of Rare Diseases</i> , 2018 , 13, 120	4.2	41
134	Variants in EXOSC9 Disrupt the RNA Exosome and Result in Cerebellar Atrophy with Spinal Motor Neuronopathy. <i>American Journal of Human Genetics</i> , 2018 , 102, 858-873	11	37
133	Two patients with MIRAGE syndrome lacking haematological features: role of somatic second-site reversion SAMD9 mutations. <i>Journal of Medical Genetics</i> , 2018 , 55, 81-85	5.8	38
132	Aicardi-Goutières syndrome with muscle involvement in early infancy. <i>Neuropathology and Applied Neurobiology</i> , 2018 , 44, 737-742	5.2	1
131	Autophagic vacuolar myopathy is a common feature of CLN3 disease. <i>Annals of Clinical and Translational Neurology</i> , 2018 , 5, 1385-1393	5.3	6
130	Transparent Danionella translucida as a genetically tractable vertebrate brain model. <i>Nature Methods</i> , 2018 , 15, 977-983	21.6	37
129	Cytoplasmic body myopathy revisited. <i>Neuromuscular Disorders</i> , 2018 , 28, 969-971	2.9	1
128	Human iPSC-Derived Neural Progenitors Are an Effective Drug Discovery Model for Neurological mtDNA Disorders. <i>Cell Stem Cell</i> , 2017 , 20, 659-674.e9	18	84
127	A novel mutation in two Turkish families associated with cerebral atrophy, global retardation, scoliosis, achalasia and alacrima. <i>Journal of Medical Genetics</i> , 2017 , 54, 176-185	5.8	34
126	Myopathology in the times of modern genetics. <i>Neuropathology and Applied Neurobiology</i> , 2017 , 43, 44-61	6.12	10
125	CoQ deficiency causes disruption of mitochondrial sulfide oxidation, a new pathomechanism associated with this syndrome. <i>EMBO Molecular Medicine</i> , 2017 , 9, 78-95	12	47
124	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. <i>American Journal of Human Genetics</i> , 2017 , 101, 833-843	11	37
123	A homozygous PIGO mutation associated with severe infantile epileptic encephalopathy and corpus callosum hypoplasia, but normal alkaline phosphatase levels. <i>Metabolic Brain Disease</i> , 2017 , 32, 2131-2137	3.9	6
122	International Workshop:: Outcome measures and clinical trial readiness in primary mitochondrial myopathies in children and adults. Consensus recommendations. 16-18 November 2016, Rome, Italy. <i>Neuromuscular Disorders</i> , 2017 , 27, 1126-1137	2.9	42
121	BMP signaling regulates satellite cell-dependent postnatal muscle growth. <i>Development (Cambridge)</i> , 2017 , 144, 2737-2747	6.6	25
120	Recessive mutation in EXOSC3 associates with mitochondrial dysfunction and pontocerebellar hypoplasia. <i>Mitochondrion</i> , 2017 , 37, 46-54	4.9	17
119	A recessive mutation in beta-IV-spectrin (SPTBN4) associates with congenital myopathy, neuropathy, and central deafness. <i>Human Genetics</i> , 2017 , 136, 903-910	6.3	32

118	Klüver-Bucy syndrome associated with a recessive variant in HGSNAT in two siblings with Mucopolysaccharidosis type IIIC (Sanfilippo C). <i>European Journal of Human Genetics</i> , 2017 , 25, 253-256	5.3	3
117	Coenzyme Q deficiency causes impairment of the sulfide oxidation pathway. <i>EMBO Molecular Medicine</i> , 2017 , 9, 96-111	12	44
116	Motor Function in Pediatric ALL Survivors after Chemotherapy-Only. <i>Neuropediatrics</i> , 2017 , 48, S1-S45	1.6	
115	Characterization of a Dmd (EGFP) reporter mouse as a tool to investigate dystrophin expression. <i>Skeletal Muscle</i> , 2016 , 6, 25	5.1	14
114	A movement disorder with dystonia and ataxia caused by a mutation in the HIBCH gene. <i>Movement Disorders</i> , 2016 , 31, 1733-1739	7	21
113	MORC2 mutation causes severe spinal muscular atrophy-phenotype, cerebellar atrophy, and diaphragmatic paralysis. <i>Brain</i> , 2016 , 139, e70	11.2	20
112	Mutations in Subunits of the Activating Signal Cointegrator 1 Complex Are Associated with Prenatal Spinal Muscular Atrophy and Congenital Bone Fractures. <i>American Journal of Human Genetics</i> , 2016 , 98, 473-489	11	42
111	Nemaline body myopathy caused by a novel mutation in troponin T1 (TNNT1). <i>Muscle and Nerve</i> , 2016 , 53, 564-9	3.4	28
110	BRAT1 mutations are associated with infantile epileptic encephalopathy, mitochondrial dysfunction, and survival into childhood. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2274-81	2.5	18
109	Altered RNA metabolism due to a homozygous RBM7 mutation in a patient with spinal motor neuropathy. <i>Human Molecular Genetics</i> , 2016 , 25, 2985-2996	5.6	27
108	A systematic, large-scale comparison of transcription factor binding site models. <i>BMC Genomics</i> , 2016 , 17, 388	4.5	10
107	Kyphoscoliosis peptidase (KY) mutation causes a novel congenital myopathy with core targetoid defects. <i>Acta Neuropathologica</i> , 2016 , 132, 475-8	14.3	11
106	Leukodystrophy with multiple beaded periventricular cysts: unusual cranial MRI results in Canavan disease. <i>Journal of Inherited Metabolic Disease</i> , 2015 , 38, 983-4	5.4	5
105	TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. <i>American Journal of Human Genetics</i> , 2015 , 97, 319-28	11	62
104	Recessive DEAF1 mutation associates with autism, intellectual disability, basal ganglia dysfunction and epilepsy. <i>Journal of Medical Genetics</i> , 2015 , 52, 607-11	5.8	18
103	Cavin 1 function does not follow caveolar morphology. <i>American Journal of Physiology - Cell Physiology</i> , 2015 , 308, C1023-30	5.4	2
102	Clinical application of whole exome sequencing reveals a novel compound heterozygous TK2-mutation in two brothers with rapidly progressive combined muscle-brain atrophy, axonal neuropathy, and status epilepticus. <i>Mitochondrion</i> , 2015 , 20, 1-6	4.9	15
101	Deficiency of ECHS1 causes mitochondrial encephalopathy with cardiac involvement. <i>Annals of Clinical and Translational Neurology</i> , 2015 , 2, 492-509	5.3	69

100	Recessive truncating IGHMBP2 mutations presenting as axonal sensorimotor neuropathy. <i>Neurology</i> , 2015 , 84, 523-31	6.5	15
99	Recessive REEP1 mutation is associated with congenital axonal neuropathy and diaphragmatic palsy. <i>Neurology: Genetics</i> , 2015 , 1, e32	3.8	16
98	Mitochondriale Erkrankungen 2015 , A13.1-A13.5		
97	Dynamics of myosin degradation in intensive care unit-acquired weakness during severe critical illness. <i>Intensive Care Medicine</i> , 2014 , 40, 528-38	14.5	85
96	A novel frameshift mutation of C19ORF12 causes NBIA4 with cerebellar atrophy and manifests with severe peripheral motor axonal neuropathy. <i>Clinical Genetics</i> , 2014 , 85, 290-2	4	9
95	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. <i>Molecular Genetics and Metabolism</i> , 2014 , 111, 342-352	3.7	58
94	Infant botulism: is there an association with thiamine deficiency?. <i>Pediatrics</i> , 2014 , 134, e1436-40	7.4	8
93	Myostatin is a key mediator between energy metabolism and endurance capacity of skeletal muscle. <i>American Journal of Physiology - Regulatory Integrative and Comparative Physiology</i> , 2014 , 307, R444-54	3.2	50
92	Blockade of ActRIIB signaling triggers muscle fatigability and metabolic myopathy. <i>Molecular Therapy</i> , 2014 , 22, 1423-1433	11.7	54
91	POMK mutation in a family with congenital muscular dystrophy with merosin deficiency, hypomyelination, mild hearing deficit and intellectual disability. <i>Journal of Medical Genetics</i> , 2014 , 51, 275-82	5.8	40
90	Comprehensive genotyping and clinical characterisation reveal 27 novel NKX2-1 mutations and expand the phenotypic spectrum. <i>Journal of Medical Genetics</i> , 2014 , 51, 375-87	5.8	60
89	Potassium channel KIR4.1-specific antibodies in children with acquired demyelinating CNS disease. <i>Neurology</i> , 2014 , 82, 470-3	6.5	41
88	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. <i>Nature Communications</i> , 2014 , 5, 4287	17.4	80
87	KIF1C mutations in two families with hereditary spastic paraparesis and cerebellar dysfunction. <i>Journal of Medical Genetics</i> , 2014 , 51, 137-42	5.8	46
86	Identifying dynamic membrane structures with atomic-force microscopy and confocal imaging. <i>Microscopy and Microanalysis</i> , 2014 , 20, 514-20	0.5	7
85	MutationTaster2: mutation prediction for the deep-sequencing age. <i>Nature Methods</i> , 2014 , 11, 361-2	21.6	2455
84	CARBON Dioxide for the treatment of Febrile seizures: rationale, feasibility, and design of the CARDIF-study. <i>Journal of Translational Medicine</i> , 2013 , 11, 157	8.5	11
83	Quantitative and qualitative 2D electrophoretic analysis of differentially expressed mitochondrial proteins from five mouse organs. <i>Proteomics</i> , 2013 , 13, 179-95	4.8	7

82	ZC4H2 mutations are associated with arthrogryposis multiplex congenita and intellectual disability through impairment of central and peripheral synaptic plasticity. <i>American Journal of Human Genetics</i> , 2013 , 92, 681-95	11	54
81	Variants in CPA1 are strongly associated with early onset chronic pancreatitis. <i>Nature Genetics</i> , 2013 , 45, 1216-20	36.3	192
80	CNVinspector: a web-based tool for the interactive evaluation of copy number variations in single patients and in cohorts. <i>Journal of Medical Genetics</i> , 2013 , 50, 529-33	5.8	3
79	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. <i>Journal of Medical Genetics</i> , 2012 , 49, 277-83	5.8	145
78	Combined effect of AAV-U7-induced dystrophin exon skipping and soluble activin Type IIB receptor in mdx mice. <i>Human Gene Therapy</i> , 2012 , 23, 1269-79	4.8	25
77	Das Deutsche Netzwerk für mitochondriale Erkrankungen (mitoNET). <i>Medizinische Genetik</i> , 2012 , 24, 193-199	0.5	2
76	The natural course of infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). <i>Pediatrics</i> , 2012 , 129, e148-56	7.4	43
75	HomozygosityMapper2012--bridging the gap between homozygosity mapping and deep sequencing. <i>Nucleic Acids Research</i> , 2012 , 40, W516-20	20.1	52
74	Region-specific expression of mitochondrial complex I genes during murine brain development. <i>PLoS ONE</i> , 2011 , 6, e18897	3.7	20
73	Recurrent stroke due to a novel voltage sensor mutation in Cav2.1 responds to verapamil. <i>Stroke</i> , 2011 , 42, e14-7	6.7	36
72	NOA1 is an essential GTPase required for mitochondrial protein synthesis. <i>Molecular Biology of the Cell</i> , 2011 , 22, 1-11	3.5	44
71	Mutations in MEGF10, a regulator of satellite cell myogenesis, cause early onset myopathy, areflexia, respiratory distress and dysphagia (EMARDD). <i>Nature Genetics</i> , 2011 , 43, 1189-92	36.3	71
70	Systematic comparison of three methods for fragmentation of long-range PCR products for next generation sequencing. <i>PLoS ONE</i> , 2011 , 6, e28240	3.7	87
69	Reply: Impaired mitochondrial function abolishes gamma oscillations in the hippocampus through an effect on fast-spiking interneurons. <i>Brain</i> , 2011 , 134, e181-e181	11.2	
68	Gamma oscillations in the hippocampus require high complex I gene expression and strong functional performance of mitochondria. <i>Brain</i> , 2011 , 134, 345-58	11.2	121
67	MutationTaster evaluates disease-causing potential of sequence alterations. <i>Nature Methods</i> , 2010 , 7, 575-6	21.6	2091
66	Treatment of CoQ(10) deficient fibroblasts with ubiquinone, CoQ analogs, and vitamin C: time- and compound-dependent effects. <i>PLoS ONE</i> , 2010 , 5, e11897	3.7	82
65	Regionalized pathology correlates with augmentation of mtDNA copy numbers in a patient with myoclonic epilepsy with ragged-red fibers (MERRF-syndrome). <i>PLoS ONE</i> , 2010 , 5, e13513	3.7	19

64	Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ10 deficiency. <i>FASEB Journal</i> , 2010 , 24, 3733-43	0.9	117
63	Fatal cardiac arrhythmia and long-QT syndrome in a new form of congenital generalized lipodystrophy with muscle rippling (CGL4) due to PTRF-CAVIN mutations. <i>PLoS Genetics</i> , 2010 , 6, e1000874	6	178
62	HomozygosityMapper--an interactive approach to homozygosity mapping. <i>Nucleic Acids Research</i> , 2009 , 37, W593-9	20.1	289
61	IGHMBP2 is a ribosome-associated helicase inactive in the neuromuscular disorder distal SMA type 1 (DSMA1). <i>Human Molecular Genetics</i> , 2009 , 18, 1288-300	5.6	69
60	Clinical variability in distal spinal muscular atrophy type 1 (DSMA1): determination of steady-state IGHMBP2 protein levels in five patients with infantile and juvenile disease. <i>Journal of Molecular Medicine</i> , 2009 , 87, 31-41	5.5	31
59	Mutations in PYCR1 cause cutis laxa with progeroid features. <i>Nature Genetics</i> , 2009 , 41, 1016-21	36.3	178
58	Protracted course of juvenile ceroid lipofuscinosis associated with a novel CLN3 mutation (p.Y199X). <i>Clinical Genetics</i> , 2009 , 76, 38-45	4	21
57	Synaptic PRG-1 modulates excitatory transmission via lipid phosphate-mediated signaling. <i>Cell</i> , 2009 , 138, 1222-35	56.2	100
56	Comparative analysis of uncoupling protein 4 distribution in various tissues under physiological conditions and during development. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , 2009 , 1788, 2309-19	3.8	50
55	Familial glucocorticoid deficiency type 1 due to a novel compound heterozygous MC2R mutation. <i>Hormone Research in Paediatrics</i> , 2008 , 69, 363-8	3.3	8
54	Respiratory chain dysfunction and oxidative stress correlate with severity of primary CoQ10 deficiency. <i>FASEB Journal</i> , 2008 , 22, 1874-85	0.9	114
53	Spinal muscular atrophy with respiratory distress type 1 (SMARD1). <i>Journal of Child Neurology</i> , 2008 , 23, 199-204	2.5	46
52	Muscle 3243A-->G mutation load and capacity of the mitochondrial energy-generating system. <i>Annals of Neurology</i> , 2008 , 63, 473-81	9.4	23
51	GeneDistiller--distilling candidate genes from linkage intervals. <i>PLoS ONE</i> , 2008 , 3, e3874	3.7	88
50	Prenatal manifestation of pancytopenia in Pearson marrow-pancreas syndrome caused by a mitochondrial DNA deletion. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 285-8	2.5	3
49	Clinical and mutational profile in spinal muscular atrophy with respiratory distress (SMARD): defining novel phenotypes through hierarchical cluster analysis. <i>Human Mutation</i> , 2007 , 28, 808-15	4.7	55
48	Detection of novel NF1 mutations and rapid mutation prescreening with Pyrosequencing. <i>Electrophoresis</i> , 2007 , 28, 4295-301	3.6	8
47	Tandem duplication of DMD exon 18 associated with epilepsy, macroglossia, and endocrinologic abnormalities. <i>Muscle and Nerve</i> , 2007 , 35, 396-401	3.4	7

46	De novo double mutation in PAX6 and mtDNA tRNA(Lys) associated with atypical aniridia and mitochondrial disease. <i>Journal of Molecular Medicine</i> , 2007 , 85, 163-8	5.5	9
45	Improved glucose metabolism in mice lacking alpha-tocopherol transfer protein. <i>European Journal of Nutrition</i> , 2007 , 46, 397-405	5.2	12
44	Lack of myostatin results in excessive muscle growth but impaired force generation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007 , 104, 1835-40	11.5	293
43	Muscle and nerve pathology in Dunnigan familial partial lipodystrophy. <i>Neurology</i> , 2007 , 68, 677-83	6.5	24
42	Analysis of mitochondrial DNA in discordant monozygotic twins with neurofibromatosis type 1. <i>Twin Research and Human Genetics</i> , 2007 , 10, 486-95	2.2	24
41	Corrigendum to Mammalian mitochondrial nitric oxide synthase: Characterization of a novel candidate [FEBS Lett. 580 (2006) 455-62]. <i>FEBS Letters</i> , 2007 , 581, 2072-2073	3.8	
40	The spectrum of WRN mutations in Werner syndrome patients. <i>Human Mutation</i> , 2006 , 27, 558-67	4.7	172
39	Genotypic and phenotypic spectrum of PANK2 mutations in patients with neurodegeneration with brain iron accumulation. <i>Annals of Neurology</i> , 2006 , 59, 248-56	9.4	158
38	Identification of small non-coding RNAs from mitochondria and chloroplasts. <i>Nucleic Acids Research</i> , 2006 , 34, 3842-52	20.1	144
37	Leigh syndrome with nephropathy and CoQ10 deficiency due to decaprenyl diphosphate synthase subunit 2 (PDSS2) mutations. <i>American Journal of Human Genetics</i> , 2006 , 79, 1125-9	11	324
36	Mammalian mitochondrial nitric oxide synthase: characterization of a novel candidate. <i>FEBS Letters</i> , 2006 , 580, 455-62	3.8	37
35	A new mutation of IGHMBP2 gene. <i>Pediatric Neurology</i> , 2006 , 34, 168	2.9	5
34	Genetic deafness in a preterm infant with a critical postnatal course. <i>Pediatric Critical Care Medicine</i> , 2006 , 7, 270-2	3	3
33	Increased mRNA expression of tissue inhibitors of metalloproteinase-1 and -2 in Duchenne muscular dystrophy. <i>Acta Neuropathologica</i> , 2005 , 109, 285-93	14.3	34
32	A two-dimensional electrophoretic map of human mitochondrial proteins from immortalized lymphoblastoid cell lines: a prerequisite to study mitochondrial disorders in patients. <i>Proteomics</i> , 2005 , 5, 2981-99	4.8	22
31	Neonatal lactic acidosis, complex I/IV deficiency, and fetal cerebral disruption. <i>Neuropediatrics</i> , 2005 , 36, 193-9	1.6	31
30	Genomic rearrangements at the IGHMBP2 gene locus in two patients with SMARD1. <i>Human Genetics</i> , 2004 , 115, 319-26	6.3	28
29	Localization of alpha-tocopherol transfer protein in trophoblast, fetal capillaries' endothelium and amnion epithelium of human term placenta. <i>Free Radical Research</i> , 2004 , 38, 413-20	4	40

28	Myostatin mutation associated with gross muscle hypertrophy in a child. <i>New England Journal of Medicine</i> , 2004 , 350, 2682-8	59.2	1044
27	Mutations in the gene encoding gap junction protein alpha 12 (connexin 46.6) cause Pelizaeus-Merzbacher-like disease. <i>American Journal of Human Genetics</i> , 2004 , 75, 251-60	11	227
26	Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). <i>Annals of Neurology</i> , 2003 , 54, 719-24	9.4	112
25	Septo-optic dysplasia associated with a new mitochondrial cytochrome b mutation. <i>Annals of Neurology</i> , 2002 , 51, 388-92	9.4	69
24	New Nuclear Encoded Mitochondrial Mutation Illustrates Pitfalls in Prenatal Diagnosis by Biochemical Methods. <i>Clinical Chemistry</i> , 2002 , 48, 772-775	5.5	15
23	New nuclear encoded mitochondrial mutation illustrates pitfalls in prenatal diagnosis by biochemical methods. <i>Clinical Chemistry</i> , 2002 , 48, 772-5	5.5	1
22	Mutations in the gene encoding immunoglobulin mu-binding protein 2 cause spinal muscular atrophy with respiratory distress type 1. <i>Nature Genetics</i> , 2001 , 29, 75-7	36.3	272
21	Multiple origins of the mtDNA 7472insC mutation associated with hearing loss and neurological dysfunction. <i>European Journal of Human Genetics</i> , 2001 , 9, 385-7	5.3	17
20	An economic method for the fluorescent labeling of PCR fragments. <i>Nature Biotechnology</i> , 2000 , 18, 233-4	44.5	2709
19	Ataxia with vitamin E deficiency: biochemical effects of malcompliance with vitamin E therapy. <i>Neurology</i> , 2000 , 55, 1584-6	6.5	12
18	A rare variant of Guillain-Barré syndrome with acute motor axonal neuropathy (AMAN) in a Caucasian boy. <i>Neuropediatrics</i> , 2000 , 31, 162-3	1.6	1
17	Heterozygous myogenic factor 6 mutation associated with myopathy and severe course of Becker muscular dystrophy. <i>Neuromuscular Disorders</i> , 2000 , 10, 572-7	2.9	45
16	Urinary α -tocopherol metabolites in α -tocopherol transfer protein-deficient patients. <i>Journal of Lipid Research</i> , 2000 , 41, 1543-1551	6.3	59
15	Urinary α -tocopherol metabolites in α -tocopherol transfer protein-deficient patients. <i>Journal of Lipid Research</i> , 2000 , 41, 1543-51	6.3	54
14	Spinal muscular atrophy-like picture, cardiomyopathy, and cytochrome c oxidase deficiency. <i>Neurology</i> , 1999 , 52, 383-6	6.5	31
13	Mutant NDUFV1 subunit of mitochondrial complex I causes leukodystrophy and myoclonic epilepsy. <i>Nature Genetics</i> , 1999 , 21, 260-1	36.3	238
12	Mutation screening of neurofibromatosis type 1 (NF1) exons 28 and 29 with single strand conformation polymorphism (SSCP): five novel mutations, one recurrent transition and two polymorphisms in a panel of 118 unrelated NF1 patients. Mutations in brief no. 229. Online. <i>Human Mutation</i> , 1999 , 13, 258	4.7	1
11	Treatment of ataxia in isolated vitamin E deficiency caused by α -tocopherol transfer protein deficiency. <i>Journal of Pediatrics</i> , 1999 , 134, 240-4	3.6	59

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9	Degenerative changes in unmyelinated nerve fibers in late-infantile neuronal ceroidlipofuscinosis. A morphometric study of conjunctival biopsy specimens. <i>Acta Neuropathologica</i> , 1998 , 95, 175-83	14.3	3
8	The first nuclear-encoded complex I mutation in a patient with Leigh syndrome. <i>American Journal of Human Genetics</i> , 1998 , 63, 1598-608	11	232
7	Cloning of the human mitochondrial 51 kDa subunit (NDUFV1) reveals a 100% antisense homology of its 3'UTR with the 5'UTR of the gamma-interferon inducible protein (IP-30) precursor: is this a link between mitochondrial myopathy and inflammation?. <i>Biochemical and Biophysical Research Communications</i> , 1998 , 245, 599-606	3.4	19
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5	Selective disactivation of neurofibromin GAP activity in neurofibromatosis type 1. <i>Human Molecular Genetics</i> , 1998 , 7, 1261-8	5.6	113
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2	Hybrid genome assembly and annotation of <i>Danio rerio</i> , a transparent fish with the smallest known vertebrate brain		2
1	Diagnosing pediatric mitochondrial disease: lessons from 2,000 exomes		3